

DIAGNOSIS OF SMITH-LEMLI-OPTIZ SYNDROME

ABSTRACT OF THE DISCLOSURE

The invention provides a non-invasive assay for the detection of a SLOS affected individual by determining the ratio of at least one of the specific SLOS steroid analytes, dehydro-estriol (8-DHE3) and a dehydro-pregnanetriol (7-DHPT) to their normal steroid counterparts estriol (E3) and pregnanetriol (PT), respectively. 8-DHE3 and 7-DHPT represent metabolites which accumulate in the blood and urine of an individual with SLOS or an individual carrying a SLOS affected fetus. The invention provides for a reliable and reproducible method of screening women for SLOS affected fetuses early on in pregnancy.

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